Current Issues in Public Health Genomics

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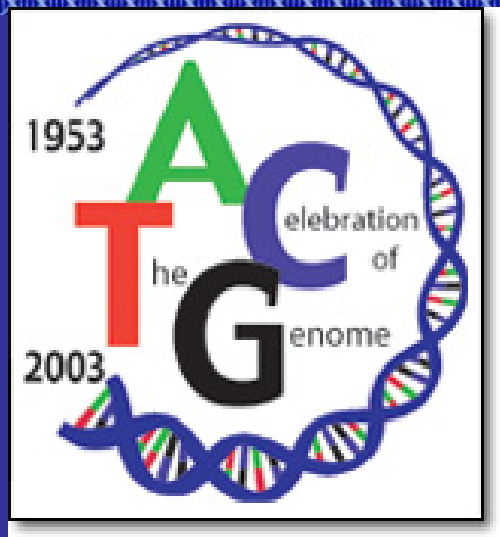
Overview

- Public Health in the Genomics Era
- Examples of CDC Public Health Genomics Building Block Initiatives
- Near Term Directions

Welcome to the Genomics Era!



Nature 2005

ARTICLES

A haplotype map of the human genome

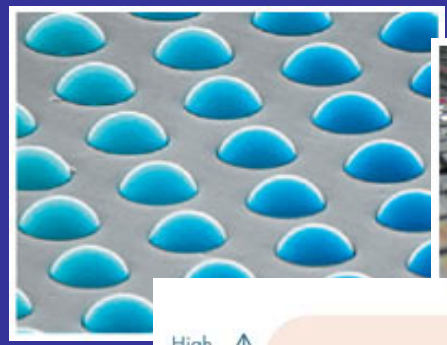
The International HapMap Consortium*

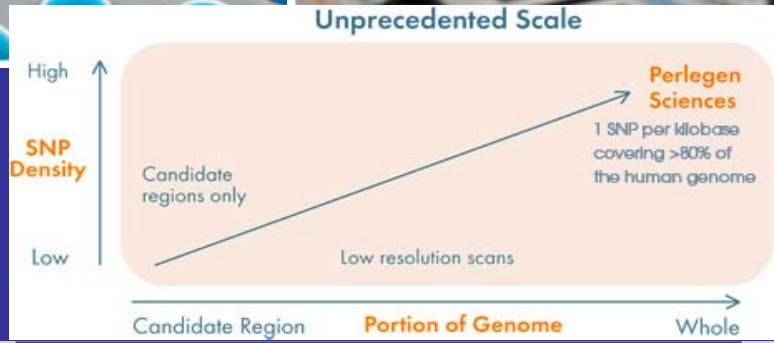
Inherited genetic variation has a critical but as yet largely uncharacterized role in human disease. Here we report a public database of common variation in the human genome: more than one million single nucleotide polymorphisms (SNPs) for which accurate and complete genotypes have been obtained in 269 DNA samples from four populations, including ten 500-kilobase regions in which essentially all information about common DNA variation has been extracted. These data document the generality of recombination hotspots, a block-like structure of linkage disequilibrium and low haplotype diversity, leading to substantial correlations of SNPs with many of their neighbours. We show how the HapMap resource can guide the design and analysis of genetic association studies, shed light on structural variation and recombination, and identify loci that may have been subject to natural selection during human evolution.

“The SNPs are down: genotyping for the rest of us”

- “Human genotyping has never been hotter, and a sophisticated set of array-based tools now simplifies the process dramatically, facilitating everything from small basic research studies to complex genetic epidemiology.”

A. Dove. Nat Methods 2005;2, 989 - 994





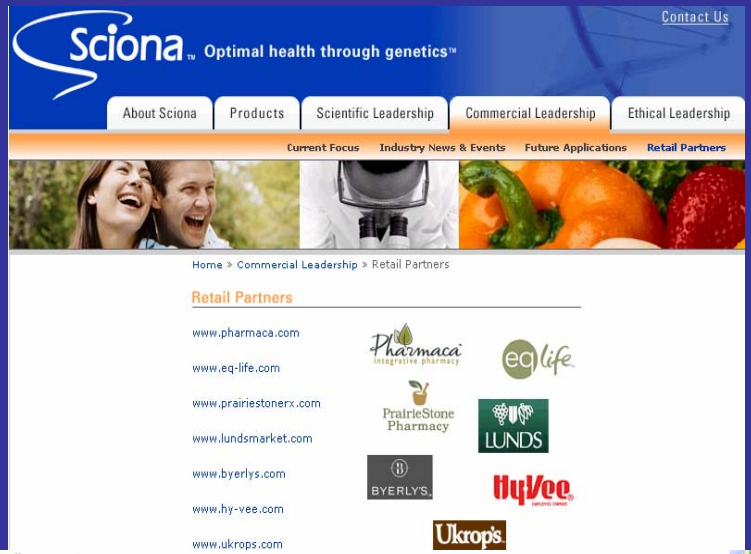
"Gene Increases Diabetes Risk, Scientists Find"

(New York Times, January 16, 2005)

- **"Scientists have discovered a variant gene that leads to a sizeable extra risk of Type 2 Diabetes and is carried by more than a third of Americans…An immediate practical consequence of the discovery would be to develop a diagnostic test to identify people who carry the variant gene. If people knew of their extra risk, they would have an incentive to stay thin and exercise."**

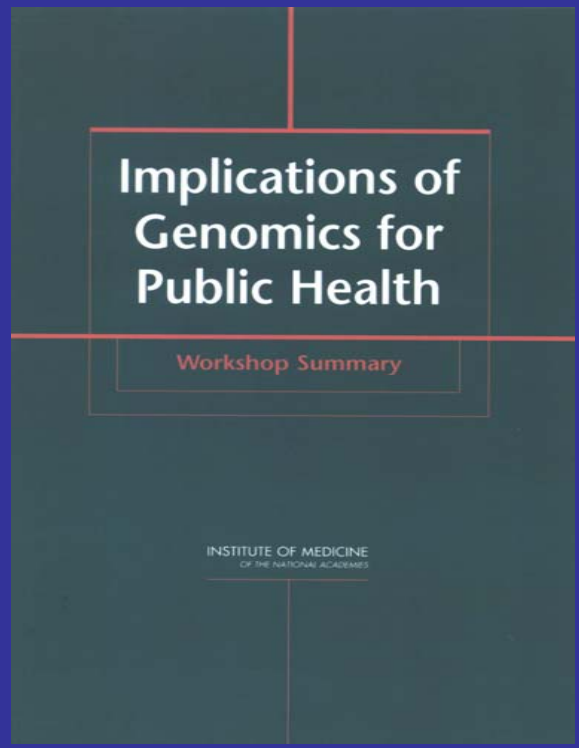
Grant SFA et al. Nat Genet Jan 2006

“Genetic Testing for Sale”

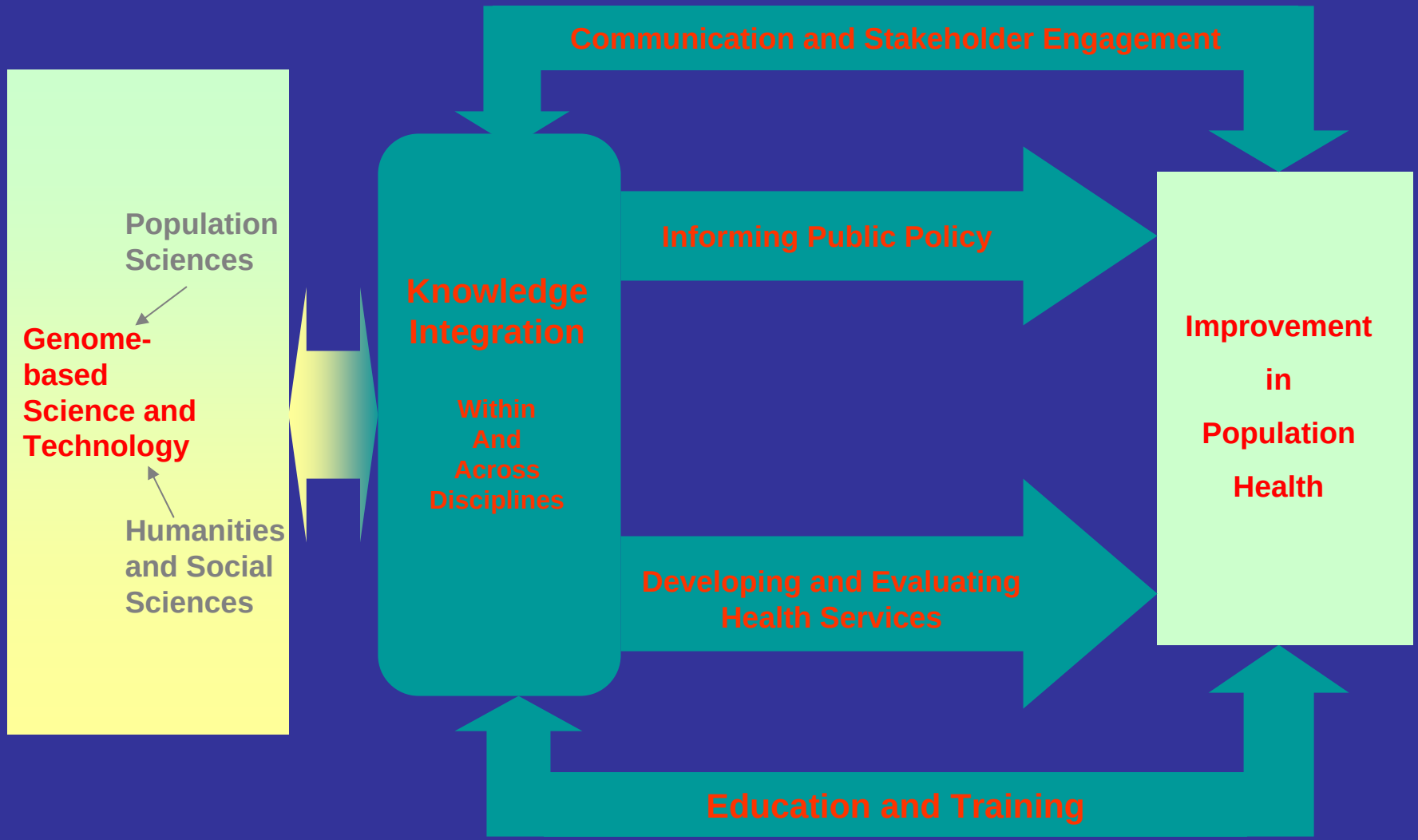


What is “Public Health Genomics”? (Institute of Medicine, 2005)

- **“An emerging field that assesses the impact of genes and their interaction with behavior, diet and the environment on the population’s health”**



The Public Health Genomics Enterprise



(The Bellagio Working Group, 2006)

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Public Health Genomics at CDC

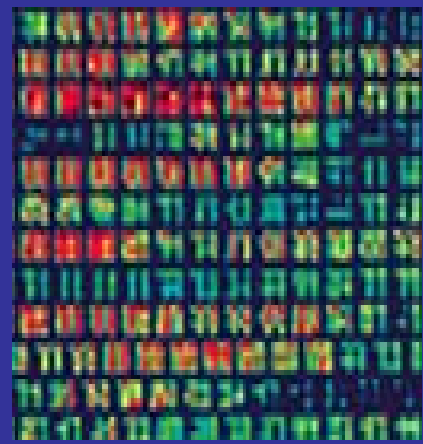
Examples of Building Block Initiatives

- Evaluation of Genomic Applications in Practice and Prevention (EGAPP)
- Family History Public Health Initiative
- National Profile of Genetic Variation
- Integrating Genomics into Public Health Investigations and Preparedness
- Public Health Genomics Response Capacity

Evaluation of Genomic Applications in Practice and Prevention (EGAPP)

Benefit: Build the Evidence Base

- Independent panel to evaluate genetic tests and other genomic applications
- Provide evidence base for practitioners and protect public from harm
- CDC accepts key leadership role recommended by many groups
- Extends CDC efforts in newborn screening and genetic testing quality assurance



Family History Public Health Initiative

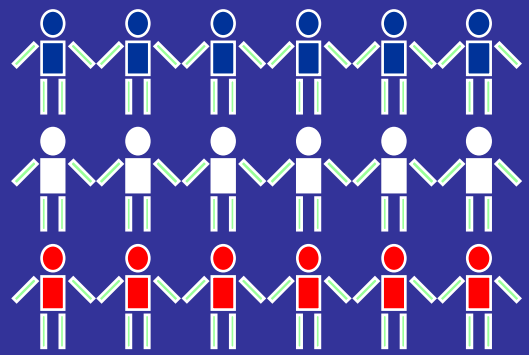
Benefit: Tools for Prevention

- Family history captures shared genes, behaviors, and environment
- Use to target screening, prevention
- CDC tool (6 diseases) & validation study in collaboration with CDC, NIH, academia
- Partnership with Surgeon General

National Profile of Genetic Variation

Benefit: Population Data for Health Impact

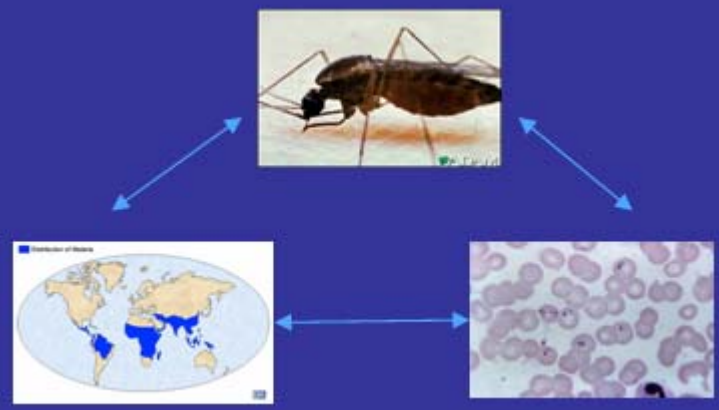
- NHANES: Representative sample of U.S. population with rich clinical, laboratory and exposure data
- Needed for estimating numbers of people at risk and who could benefit from interventions
- CDC working group projects:
  - Phase I: 100-200 variants
  - Phase II: 300,000-500,000 variants



Genomics in Public Health Investigations

Benefit: Prevent and Control Disease in Communities

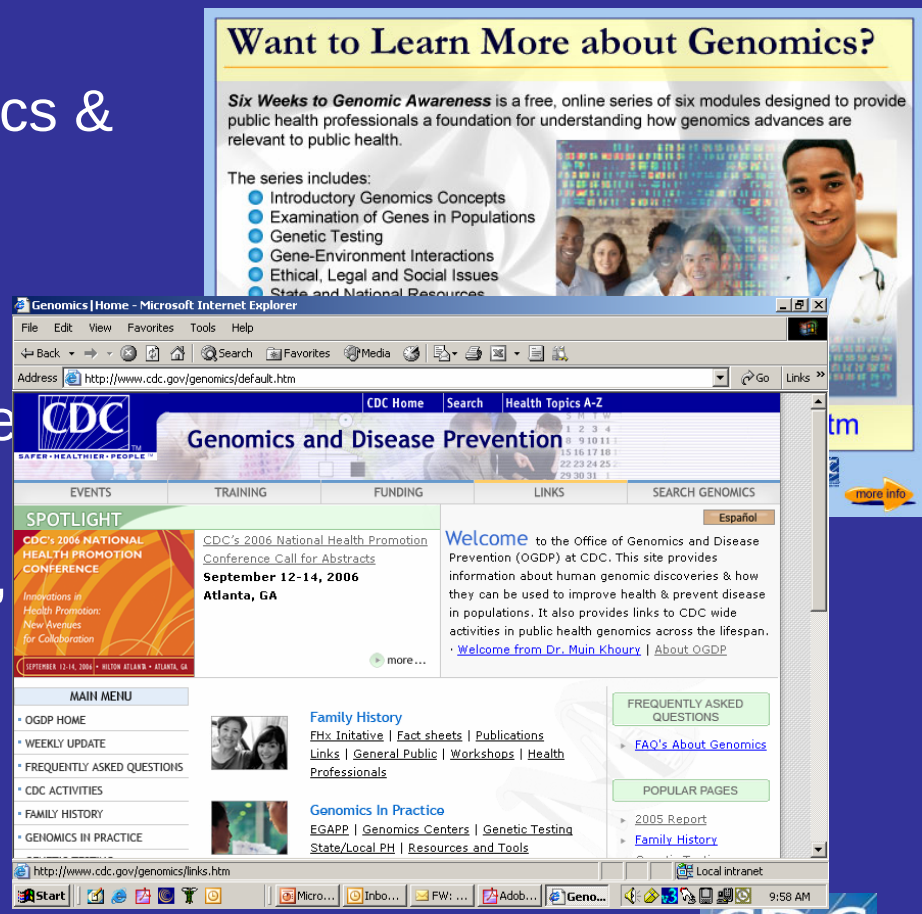
- Pathogen genomics a key public health tool
- Human genomics: susceptibility, vaccine and drug response, adverse effects
- Identify environmental factors for intervention



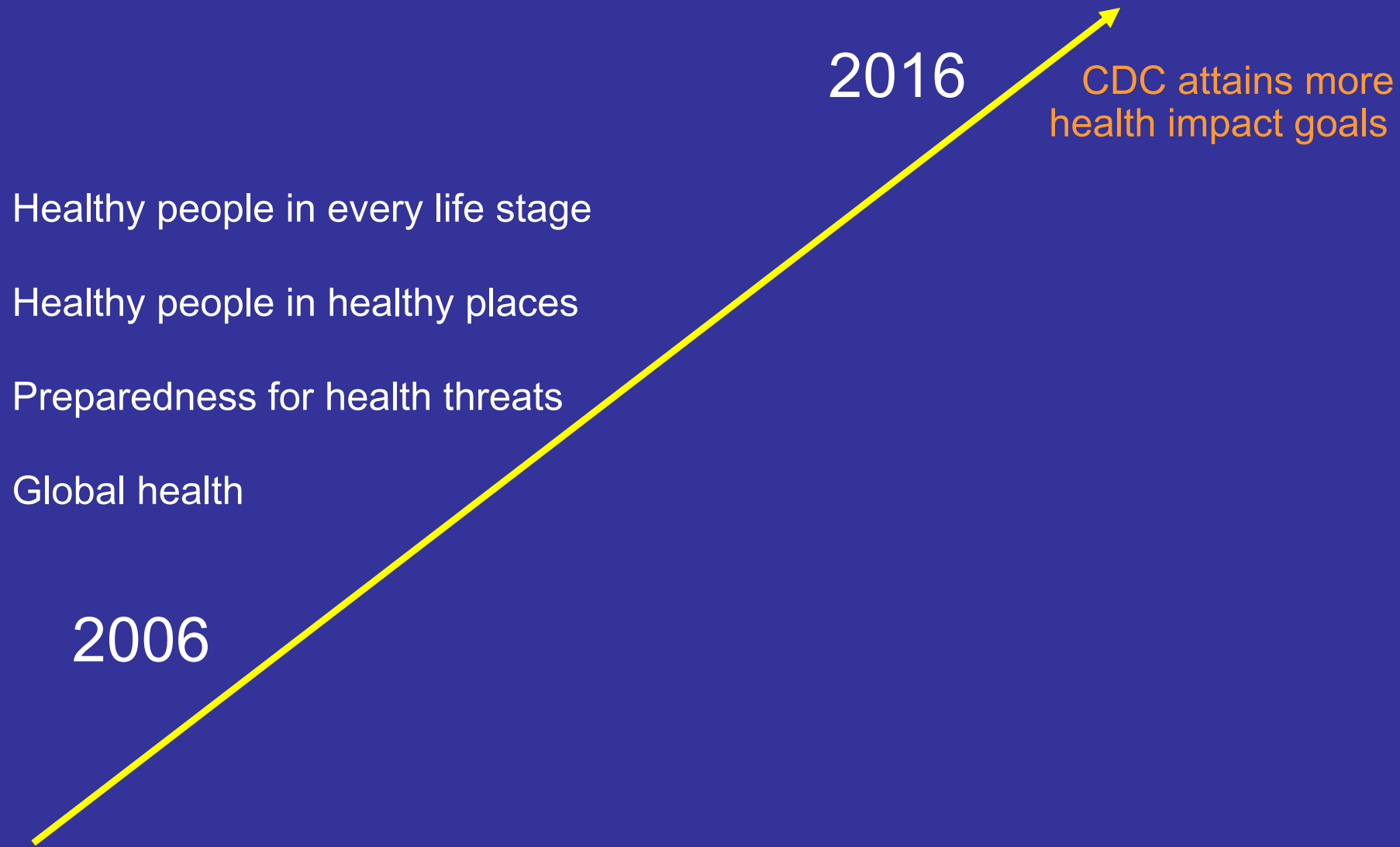
Public Health Genomics Response Capacity

Benefit: Prepare public health capacity & workforce

- Centers for excellence in genomics & public health
- Integrating genomics into state programs
- Public health genomics workforce competencies
- Reports, website, online courses, weekly update



Public Health Genomics: a Vision for the Future



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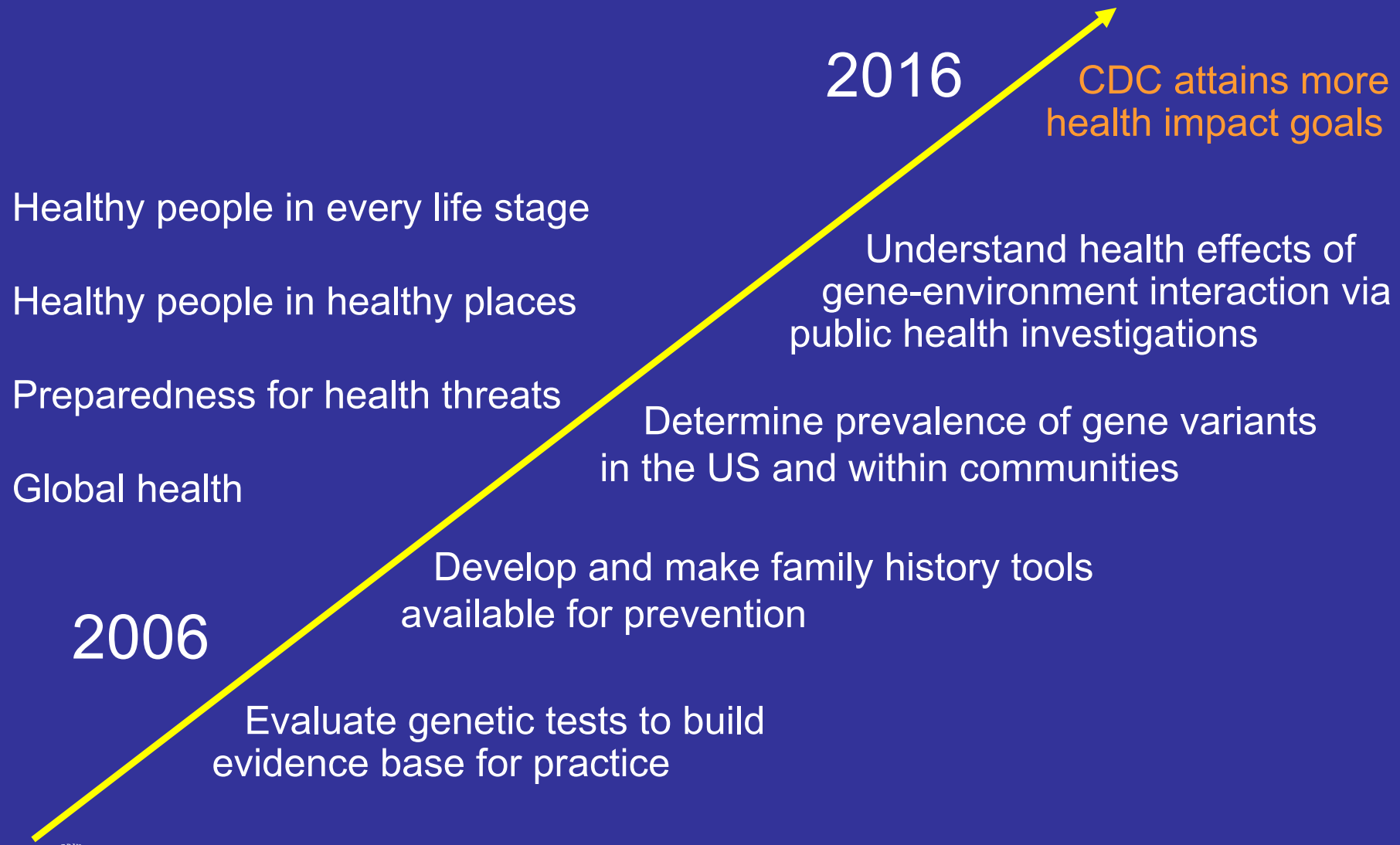

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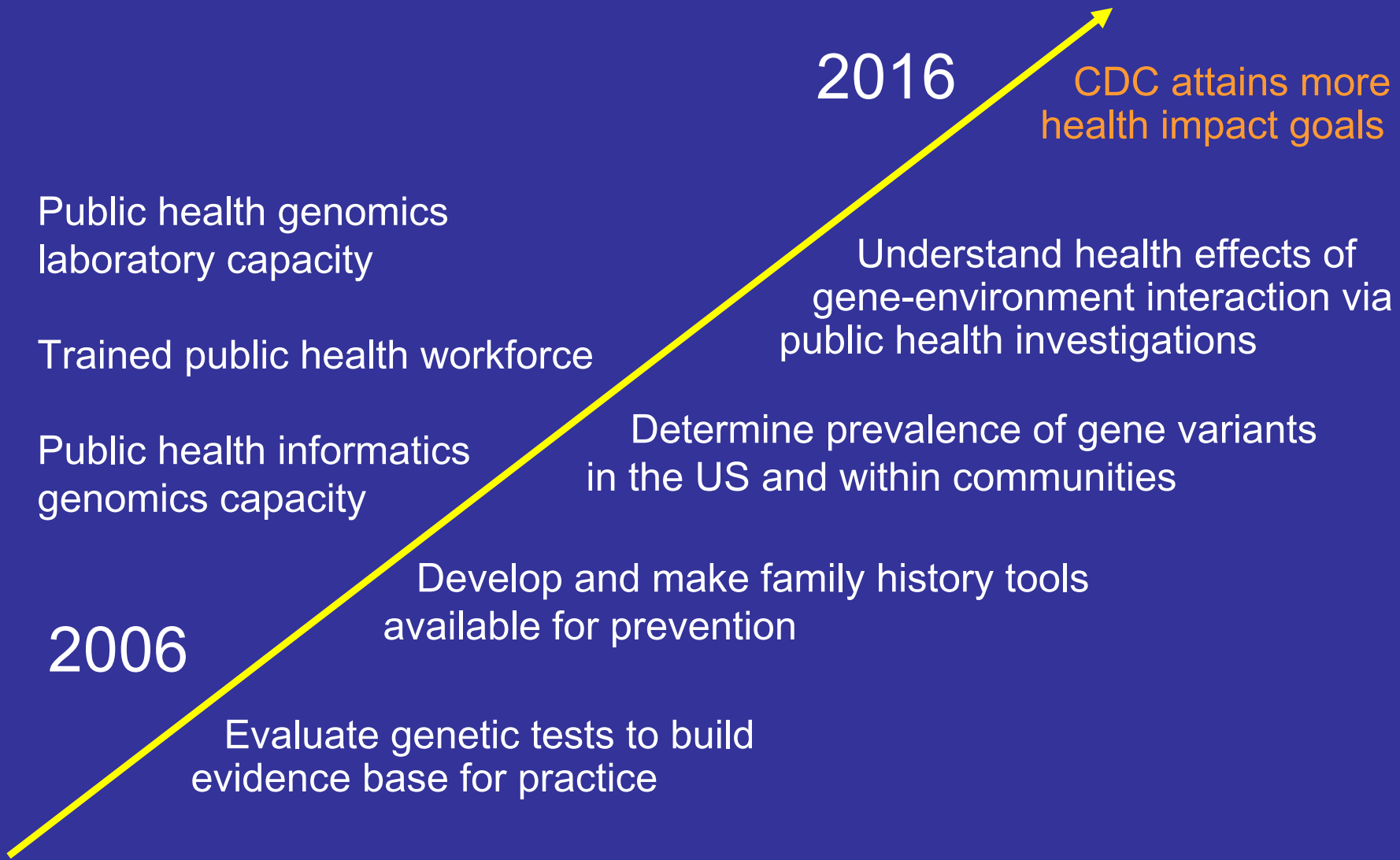

Near Term Directions in Public Health Genomics at CDC

- Enhancing networking and collaborations
- Developing CDC goals oriented initiatives
- Making seed funding available
- Emphasizing career development